1/13

SEQUENCE LISTING

<110>	The Hospital for Sick Children	
S	Scherer, Stephen W.	
ı	Minassian, Berge A.	
<120>	Novel Lafora's Disease Gene	
<130>	9962-58	
<150>	US 60/491,968	
<151>	2003-08-04	
<160>	5	
<170>	PatentIn version 3.1	
<210>	1	•
<211>	2120	
<212>	DNA	
<213>	Homo sapiens	
<400>	1 ggccg aagcctcgga gagcgggcca gcgctgcatg agctcatgcg cgaggcggag	60
	cctgc tcgagtgcaa ggtgtgcttt gagaagtttg gccaccggca gcagcggcgc	120
	caacc tgtcctgcgg ccacgtggtc tgcctggcct gcgtggccgc cctggcgcac	180
	cactc tggccctcga gtgcccattc tgcaggcgag cttgccgggg ctgcgacacc	240
	ctgcc tgccggtgct gcacctcata gagctcctgg gctcagcgct tcgccagtcc	300
	egece ategegeege ecceagegee eceggageee teacetgeea ceacacette	360

ggcggctggg ggaccctggt caaccccacc ggactggcgc tttgtcccaa gacggggcgt

420

2/13

gtcgtggtgg tgcacgacgg caggag	ggcgt gtcaagattt	ttgactcagg	gggaggatgc	480
gcgcatcagt ttggagagaa ggggga	acgct gcccaagaca	ttaggtaccc	tgtggatgtc	540
accatcacca acgactgcca tgtggt	ttgtc actgacgccg	gcgatcgctc	catcaaagtg	600
tttgattttt ttggccagat caagct	ttgtc attggaggcc	aattctcctt	accttggggt	660
gtggagacca cccctcagaa tggga	ttgtg gtaactgatg	cggaggcagg	gtccctgcac	720
ctcctggacg tcgacttcgc ggaagg	gggtc cttcggagaa	ctgaaaggtt	gcaagctcat	780
ctgtgcaatc cccgaggggt ggcag	tgtct tggctcaccg	gggccattgc	ggtcctggag	840
cacccctgg ccctggggac tgggg	tttgc agcaccaggg	tgaaagtgtt	tagctcaagt	900
atgcagcttg tcggccaagt ggata	ccttt gggctgagcc	tctactttcc	ctccaaaata	960
actgcctccg ctgtgacctt tgatc	accag ggaaatgtga	ttgttgcaga	tacatctggt	1020
ccagctatcc tttgcttagg aaaac	ctgag gagtttccag	taccgaagcc	catggtcact	1080
catggtcttt cgcatcctgt ggctc	ttacc ttcaccaagg	agaattctct	tcttgtgctg	11:40
gacacagcat ctcattctat aaaag	tctat aaagttgact	gggggtgatg	ggctggggtg	1200
ggtccctgga atcagaagca ctagt	gctgc cattaatgaa	ttgtttaacc	ctggataagt	1260
cacttaaact catctatcca ggcag	ggata attaaaacca	tctggcagac	ttacaaagct	1320
tgggacagtt attggagatt aatct	accat ttattgaatg	catactctgt	gcaaggaaat	1380
ttgcaaatat tagcttattt aatct	gtact atccagtgag	gtaatttctt	ccccccaag	1440
atagagtcaa gctctgtcac ccagg	ctgga gtgcagaago	atgatcacag	ctcactacag	1500
tttcaacgtc ccccgctcag gtggt	ecttc cacctcagco	tcccaagtag	ctgggaccac	1560
aagtgtgcat taccacactc agcta	atttt tgtattttgg	g cagagatggg	gtttcaccat	1620
gttgcccagg ctggtctcaa actcc	tgagt tcaagcaatc	caccttcctc	ggcctcccaa	1680
agtactagga gtacaggcat agcca	cttgc tcagccataa	tttttattat	taatctcatt	1740
gtacaagtga gaaaactgag accca	gagag cttaagtgad	ttcctcgagg	tcatagttac	1800
ttactgcctt agtcccaatt tgaat	tcaat tctgattcca	a aataagttgc	gcttaaataa	1860
gacaacagat gtgggaaaaa tatgt	gaatg tgtagtgtt	g ctatgtgtac	tgtctttaca	1920
agtagctaat tattttagca caaag	gatgtg caaagaaagg	g agactttatg	gagagttcag	1980
gagaaaaagg attttgtggt ggcca	atcact ttcattcaat	ttgcgactgc	tctgatggca	2040
cattagatga agttactgtt gatco	ctgagt tacgtgaate	a agaaaaacaa	ttgaactgct	2100
tattaaaaaa gtaaacatgt				2120

3/13

<210> 2

<211> 395

<212> PRT

<213> Homo sapiens

<400> 2

Met Ala Ala Glu Ala Ser Glu Ser Gly Pro Ala Leu His Glu Leu Met 1 5 10 15

Arg Glu Ala Glu Ile Ser Leu Leu Glu Cys Lys Val Cys Phe Glu Lys 20 25 30

Phe Gly His Arg Gln Gln Arg Arg Pro Arg Asn Leu Ser Cys Gly His 35 40 45

Val Val Cys Leu Ala Cys Val Ala Ala Leu Ala His Pro Arg Thr Leu 50 55 60

Ala Leu Glu Cys Pro Phe Cys Arg Arg Ala Cys Arg Gly Cys Asp Thr 65 70 75 80

Ser Asp Cys Leu Pro Val Leu His Leu Ile Glu Leu Leu Gly Ser Ala 85 90 95

Leu Arg Gln Ser Pro Ala Ala His Arg Ala Ala Pro Ser Ala Pro Gly
100 105 110

Ala Leu Thr Cys His His Thr Phe Gly Gly Trp Gly Thr Leu Val Asn 115 120 125

Pro Thr Gly Leu Ala Leu Cys Pro Lys Thr Gly Arg Val Val Val 130 135 140

His Asp Gly Arg Arg Val Lys Ile Phe Asp Ser Gly Gly Cys 145 150 155 160

Ala His Gln Phe Gly Glu Lys Gly Asp Ala Ala Gln Asp Ile Arg Tyr 165 170 175

Pro Val Asp Val Thr Ile Thr Asn Asp Cys His Val Val Val Thr Asp 180 185 190

4/13

Ala Gly Asp Arg Ser Ile Lys Val Phe Asp Phe Phe Gly Gln Ile Lys 195 200 205

Leu Val Ile Gly Gly Gln Phe Ser Leu Pro Trp Gly Val Glu Thr Thr 210 215 220

Pro Gln Asn Gly Ile Val Val Thr Asp Ala Glu Ala Gly Ser Leu His 225 230 235 240

Leu Leu Asp Val Asp Phe Ala Glu Gly Val Leu Arg Arg Thr Glu Arg 245 250 255

Leu Gln Ala His Leu Cys Asn Pro Arg Gly Val Ala Val Ser Trp Leu 260 265 270

Thr Gly Ala Ile Ala Val Leu Glu His Pro Leu Ala Leu Gly Thr Gly 275 280 285

Val Cys Ser Thr Arg Val Lys Val Phe Ser Ser Ser Met Gln Leu Val 290 295 300

Gly Gln Val Asp Thr Phe Gly Leu Ser Leu Tyr Phe Pro Ser Lys Ile 305 310 315 320

Thr Ala Ser Ala Val Thr Phe Asp His Gln Gly Asn Val Ile Val Ala 325 330 335

Asp Thr Ser Gly Pro Ala Ile Leu Cys Leu Gly Lys Pro Glu Glu Phe 340 345 350

Pro Val Pro Lys Pro Met Val Thr His Gly Leu Ser His Pro Val Ala 355 360 365

Leu Thr Phe Thr Lys Glu Asn Ser Leu Leu Val Leu Asp Thr Ala Ser 370 375 380

His Ser Ile Lys Val Tyr Lys Val Asp Trp Gly 385 390 395

<210> 3

<211> 3008

<212> DNA

5/13

<213> Canis sp. <220> <221> CDS <222> (698)..(1897) <223> <220> <221> misc_feature <222> (2692)..(2692) <223> N=any nucleic acid <220> <221> misc_feature <222> (2748)..(2748) <223> N=any nucleic acid <220> <221> misc_feature <222> (2750)..(2750) <223> N=any nucleic acid <220> <221> misc_feature <222> (2793)..(2793) <223> N=any nucleic acid <220> <221> misc_feature <222> (2845)..(2845)

6/13

<223> N=any nucleic acid <220> <221> misc_feature <222> (2916)..(2916) <223> N=any nucleic acid <220> <221> misc_feature <222> (2918)..(2918) <223> N=any nucleic acid <220> <221> misc_feature <222> (2931)..(2931) <223> N=any nucleic acid <220> <221> misc_feature <222> (2941)..(2941) <223> N=any nucleic acid <220> <221> misc_feature <222> (2990)..(2990) <223> N=any nucleic acid <400> 3 ccccaaggcc ccccggccc ccaggcaacc ccaggccccc aggcaaccca aggccccccg 60 gccccaagcc ccccaggttc ccggccccaa gaaccaagcc ccccggcccc ccgccccag 120

7/13

cacccagca	c caagc	cccg c	ccccgc	cc caa	gcaccc	a gcccc	agcac	ccagc	cccg	180
cccagccc	c agccc	cagca c	ccagccc	cc gcc	ccagca	c ccago	cccag	caccca	agccc	240
ccgcccag	c cccag	cccc g	tecece	cc cca	gcaccc	a gcccc	agccc	cagcag	gcagc	300
acccagcag	g ggact	gcaaa g	cgtaggc	ta ccc	caggtg	g aacac	egtgt	tctag	ttttg	360
ctttgccgt	t tgcag	cctgg g	cgatcgg	gg gcc	accgct	c gagco	tgttt	cccgt	cgcgg	420
aaagcggag	c cgccc	cgccc c	gccccc	gc ctg	cctgaa	g gtcad	egggcc	tgggc	ctgcg	480
gcgcgcggt	g eggee	cgcga g	cgtccgc	tc ccg	cgccct	c cgcaç	gtcagc	gcccg	cccgc	540
ccgccgggg	g accgo	aggcc g	cggccga	ga ggc	tgcgcg	c tgcg	cccgcg	acgtc	aggcc ·	600
ccgccccgc	c ccgcc	ccgcc c	cgtgacc	gg ccc	cggccc	c ggcc	ccggcc	ccggc	cccgg	660
accgagcgg	ge geeeg	geggga g	cggcggc	gg ccg	cgcg a M	Met Gly	gcc ga Ala Gl	aa gcg lu Ala 5	gcg Ala	715
ggg agc (Gly Ser (ggg cgg 3ly Arg 10	gcg ctg Ala Leu	cgg ga Arg Gl	ig ctg Lu Leu 15	gtg cg Val Ar	gc gag g gg Glu i	gcc gag Ala Glu 20	g gtc ı Val	agc Ser	763 <u>.</u>
ttg ctc g	gag tgc Glu Cys 25	aag gtg Lys Val	tgc tt Cys Ph 30	ne Glu	agg tt Arg Pl	ne Gly	cac cgo His Aro 35	c cag g Gln	cag Gln	811
cgg cgc (Arg Arg 40	ccg cgc Pro Arg	aac cto Asn Lev	g ccc to L Pro Cy 45	As GJA	cac g [†] His Va	tg.gtg al Val 50	tgc cte Cys Le	g gcc u Ala	tgc Cys	859
gtg gcg g Val Ala s 55	gcc ctg Ala Leu	gcg cac Ala His 60	ccg cg	gg acg rg Thr	ctg go Leu A	la Leu	gag tg Glu Cy	c ccc s Pro	ttc Phe 70	907
tgc cgc Cys Arg	cgg gcc Arg Ala	tgc cgc Cys Arg 75	g ggc to	gc gac ys Asp	acc ag Thr So	gc gac er Asp	tgc ct Cys Le	g ccg u Pro 85	gtg Val	955
ctt cac Leu His	ctc ctg Leu Leu 90	gag cto	c ctg go 1 Leu G	gc tcg ly Ser 95	gcg c Ala L	tg cgc eu Arg	cca gc Pro Al 10	a Pro	gcc Ala	1003
gcc ccc Ala Pro	cgc gcc Arg Ala 105	gcc cc Ala Pr	o Arg A	cc gcc la Ala 10	ccc t Pro C	gc gcc ys Ala	ccg gg Pro Gl 115	c gcc y Ala	ctc Leu	1051
gcc tgc Ala Cys 120	cat cac His His	gcg tt Ala Ph	c gga g e Gly G 125	gc tgg ly Trp	ggg a Gly I	cc ctg hr Leu 130	gtc aa Val As	c ccc n Pro	acg Thr	1099
ggg ctg Gly Leu 135	gcg ctg Ala Leu	tgc cc Cys Pr 14	o Lys T	cc ggg hr Gly	Arg V	tc gtg Val Val	gtg gt Val Va	g cac l His	gac Asp 150	1147

8/13

ggc Gly	agg Arg	agg Arg	cgg Arg	gtc Val 155	aag Lys	atc Ile	ttt Phe	gac Asp	tcc Ser 160	ggg ggg	gga Gly	gga Gly	tgc Cys	gcc Ala 165	cat His	1195
cag Gln	ttt Phe	gga Gly	gag Glu 170	aag Lys	GJA aaa	gag Glu	gct Ala	gcc Ala 175	cag Gln	gac Asp	att Ile	agg Arg	tac Tyr 180	ccc Pro	ctg Leu	1243
gac Asp	gtc Val	gcc Ala 185	gtc Val	acc Thr	aac Asn	gac Asp	tgc Cys 190	cac His	gtg Val	gtt Val	gtc Val	acc Thr 195	gac Asp	gcc Ala	ggc	1291
gac Asp	cgc Arg 200	tcc Ser	atc Ile	aaa Lys	gtg Val	ttt Phe 205	gat Asp	ttc Phe	ttt Phe	ggc Gly	cag Gln 210	atc Ile	aag Lys	ctc Leu	gtc Val	1339
att Ile 215	gga Gly	gac Asp	cag Gln	ttt Phe	tcc Ser 220	tta Leu	cct Pro	tgg Trp	ggc	gtg Val 225	gag Glu	acc Thr	acc Thr	cct Pro	cag Gln 230	1387
aat Asn	GJÀ âãa	gtc Val	gtg Val	gta Val 235	act Thr	gac Asp	gcc Ala	gag Glu	gca Ala 240	Gly aaa	tcg Ser	ctg Leu	cac His	ctg Leu 245	ctg Leu	1435
gaa Glu	gtc Val	gac Asp	ttt Phe 250	gca Ala	gaa Glu	gga Gly	gcc Ala	ctc Leu 255	cag Gln	agg Arg	act Thr	gaa Glu	aag Lys 260	ctg Leu	caa Gln	1483
ggt Gly	cat His	ctg Leu 265	Cys	aac Asn	ccg Pro	cga Arg	ggg Gly 270	Val	gcc Ala	gtg Val	tcc Ser	tgg Trp 275	ctc Leu	act Thr	GJA āāā	1531
gcc Ala	att Ile 280	Ala	gtc Val	ctg Leu	gag Glu	cac His 285	cct Pro	ccg Pro	gly aaa	ctg Leu	ggg Gly 290	Ala	GJÀ aaa	gcg Ala	ggc Gly	1579
agc Ser 295	Thr	gcc Ala	gtg Val	aag Lys	gtg Val 300	ttc Phe	agc Ser	cca Pro	act Thr	atg Met 305	Gln	ctg Leu	atc	ggc	cag Gln 310	1627
gtg Val	gat Asp	acc	ttt Phe	Gly	ctc Leu	Ser	Leu	Phe	Phe 320	Pro	tct Ser	aga Arg	ata Ile	acc Thr 325	gcc Ala	1675
tcc Ser	gcc Ala	gtg Val	acc Thr 330	Phe	gat Asp	cac	cag Gln	ggg Gly 335	Asn	gtg Val	att Ile	gtt Val	gca Ala 340	. Asp	act Thr	1723
tct Ser	agt Ser	cag Gln 345	Ala	gto Val	cta Leu	tgc Cys	Leu 350	ı Gly	cag Gln	cct Pro	gag Glu	gaa Glu 355	Phe	cca Pro	gtc Val	1771
ctg	aag Lys 360	Pro	ato Ile	ato Ile	acc Thr	cat His 365	Gl _y	ctt Leu	tco Ser	cat His	cct Pro 370	Val	gca Ala	ctg Lev	acc Thr	1819
tto Phe 375	Thr	aag Lys	gag Glu	g aat 1 Asr	tct Ser 380	Leu	ctt Lei	gtg Val	cto Lev	gac Asp 385	Ser	gca Ala	gco Ala	cat His	tcc Ser 390	1867

9/13

gta aaa gtc ta Val Lys Val Ty	ac aag gct yr Lys Ala 395	gac tgg gg Asp Trp G	gg taa tggg ly	gtgtgg tggg	ggteet	1917
ggaactgcca cta	aatccagt t	taaccctgg	atgaattaat	cccatctctc	gaacggggat	1977
cattataact gc	ctgacaga c	cttataaagg	ttgaaggtaa	ttattaaaga	ataataatga	2037
agtctaccgt tt	attgagtt a	atgtgctccc	tgtgctagga	aactttgcaa	atattagctc	2097
agcgtgtcct ta	cagtggta c	ccagggagg	taatgcccat	cattaatccc	attttagaga	2157
tgagaaaact ga	gacccgag g	ggtttaagtg	attctctgaa	ggtcatgttt	acttactgtg	2217
acagtcacaa tg	ggaactct a	attctgactc	cccaatccct	tgctcctaag	taggataaca	2277
gatgtgagaa aa	cgacagca t	tgtgtctata	tgttgttact	gtgtgtactc	tctttacagg	2337
tagctatttc tc	ttggttgg a	acgtgcagag	aaaggagact	ttctagagag	ttcaagagga	2397
aaaagggtag tg	tgatgagc	atggacgtga	gtgtcattga	acttgctggt	tctttgatgt	2457
cacagtaggt ag	gaatgactg	tggatccttc	aactgccctt	gggaaaggta	aacatgtctg	2517
ttgggacctg ga	atgtcctcc	atcataggaa	cccaggaaat	actagttggt	tgctgcagaa	2577
aggcttgtgt gg	gacataagt	tcaaaactac	tgccgaccac	cgtacattca	cacacctcca	2637
gtgggagatg go	ctggaagac	agtcctgtga	caggtctgca	ttcatagaac	aagangccgc	2697
caccgttggt to	cacggcaga	atgagtttgc	ctgcctcttc	ataatctgtg	ncnacccgaa	2757
accettttgt ga	atagagttt	ttctctgtgc	catttnaatt	tgtcccattg	cacacactgt	2817
tttcccctaa co	cagctccct	tgatgctnag	ctagcattta	ggccactggt	aaacccctgt	2877
atacttcttg ag	gttgaagtt	aagctttgac	ccagataang	nctgctttaa	tacntgcagt	2937
cgantggacc ga	aataagggg	gaaatttcag	gtgaggtggc	: cgggttcttt	atnaaccggt	2991
tttggtttgt a						3008

<210> 4

<211> 399

<212> PRT

<213> Canis sp.

<220>

<221> misc_feature

<222> (2692)..(2692)

10/13

<223> N=any nucleic acid <220> <221> misc_feature <222> (2748)..(2748) <223> N=any nucleic acid <220> <221> misc_feature <222> (2750)..(2750) <223> N=any nucleic acid <220> <221> misc_feature <222> (2793)..(2793) <223> N=any nucleic acid <220> <221> misc_feature <222> (2845)..(2845) <223> N=any nucleic acid <220> <221> misc_feature <222> (2916)..(2916) <223> N=any nucleic acid <220> <221> misc_feature <222> (2918)..(2918) <223> N=any nucleic acid <220> <221> misc_feature <222> (2931)..(2931) <223> N=any nucleic acid

11/13

<220>

<221> misc_feature

<222> (2941)..(2941)

<223> N=any nucleic acid

<220>

<221> misc_feature

<222> (2990)..(2990)

<223> N=any nucleic acid

<400> 4

Met Gly Ala Glu Ala Ala Gly Ser Gly Arg Ala Leu Arg Glu Leu Val 1 5 10 15

Arg Glu Ala Glu Val Ser Leu Leu Glu Cys Lys Val Cys Phe Glu Arg 20 25 30

Phe Gly His Arg Gln Gln Arg Arg Pro Arg Asn Leu Pro Cys Gly His 35 40 45

Val Val Cys Leu Ala Cys Val Ala Ala Leu Ala His Pro Arg Thr Leu 50 55 60

Ala Leu Glu Cys Pro Phe Cys Arg Arg Ala Cys Arg Gly Cys Asp Thr 65 70 75 80

Ser Asp Cys Leu Pro Val Leu His Leu Leu Glu Leu Leu Gly Ser Ala 85 90 95

Leu Arg Pro Ala Pro Ala Ala Pro Arg Ala Ala Pro Arg Ala Ala Pro 100 105 110

Cys Ala Pro Gly Ala Leu Ala Cys His His Ala Phe Gly Gly Trp Gly 115 120 125

Thr Leu Val Asn Pro Thr Gly Leu Ala Leu Cys Pro Lys Thr Gly Arg 130 135 140

Val Val Val His Asp Gly Arg Arg Val Lys Ile Phe Asp Ser 145 150 155 160

Gly Gly Cys Ala His Gln Phe Gly Glu Lys Gly Glu Ala Ala Gln

12/13

175 170 165 Asp Ile Arg Tyr Pro Leu Asp Val Ala Val Thr Asn Asp Cys His Val 185 180 Val Val Thr Asp Ala Gly Asp Arg Ser Ile Lys Val Phe Asp Phe Phe 200 Gly Gln Ile Lys Leu Val Ile Gly Asp Gln Phe Ser Leu Pro Trp Gly 215 Val Glu Thr Thr Pro Gln Asn Gly Val Val Val Thr Asp Ala Glu Ala 235 Gly Ser Leu His Leu Leu Glu Val Asp Phe Ala Glu Gly Ala Leu Gln 250 Arg Thr Glu Lys Leu Gln Gly His Leu Cys Asn Pro Arg Gly Val Ala 265 Val Ser Trp Leu Thr Gly Ala Ile Ala Val Leu Glu His Pro Pro Gly 280 275 Leu Gly Ala Gly Ala Gly Ser Thr Ala Val Lys Val Phe Ser Pro Thr Met Gln Leu Ile Gly Gln Val Asp Thr Phe Gly Leu Ser Leu Phe Phe 315 310 Pro Ser Arg Ile Thr Ala Ser Ala Val Thr Phe Asp His Gln Gly Asn 335 330 325 Val Ile Val Ala Asp Thr Ser Ser Gln Ala Val Leu Cys Leu Gly Gln 350 345 Pro Glu Glu Phe Pro Val Leu Lys Pro Ile Ile Thr His Gly Leu Ser 360 His Pro Val Ala Leu Thr Phe Thr Lys Glu Asn Ser Leu Leu Val Leu 375 Asp Ser Ala Ala His Ser Val Lys Val Tyr Lys Ala Asp Trp Gly

390

385

395

13/13

<210> 5

<211> 12

<212> DNA

<213> Canis sp.

<400> 5 gccgccccc gc

12